

## Suggested Follow-up for Elevated C14:1 Tetradecenoyl Carnitine

**Possible Causes:** Elevated C14:1 is the primary marker for **very long chain acyl co-A dehydrogenase deficiency (VLCAD)**. VLCAD is a defect in fatty acid oxidation.

**Next Steps if Abnormal:** **Potential medical emergency.** See infant as soon as possible to ascertain health status. Consult pediatric metabolic specialist and initiate diagnostic evaluation and treatment as recommended. Common diagnostic studies include plasma total and free carnitines, plasma acylcarnitines and urine organic acids. For other findings, collect blood spot specimen for repeat acyl carnitine profile. A portion of the initial specimen will be sent to the Greenwood Genetic Center Laboratory for secondary testing.

**Neonatal Presentation:** Hypoketotic hypoglycemia, hepatic dysfunction, hypotonia and cardiomyopathy. Infants are at risk for metabolic decompensation/crisis.

**Emergency Treatment:** Treatment of metabolic crisis includes provision of sufficient calories (concentrated dextrose infusion with appropriate electrolytes) to correct catabolic state and biochemical abnormalities if needed.

**Standard Treatment:** Avoid fasting. Feed every four hours through the night for first several months. Fat restricted diet with use of MCT oil as fat source. May need cornstarch supplementation at bedtime to maintain blood glucose levels overnight. Carnitine supplementation if helpful.

**Advice for Family:** Provide basic information about fatty acid disorders. The handout, *When Baby Needs a Second Test for a Fatty Acid Disorder (Elevated C14:1)*, may be used for this purpose. Stress the importance of seeking immediate medical attention if the infant shows any signs of illness.

**NOTE:** Screening may also identify carriers of VLCAD.

**Internet Resources:**

<http://oregon.gov/DHS/ph/nbs/expand.shtml>

[http://web1.tch.harvard.edu/newenglandconsortium/scientists\\_physicians2.html](http://web1.tch.harvard.edu/newenglandconsortium/scientists_physicians2.html)

<http://ghr.nlm.nih.gov/condition=verylongchainacylcoenzymeadehydrogenasedeficiency>